# GENOMICS: WHY SHOULD WE CARE?

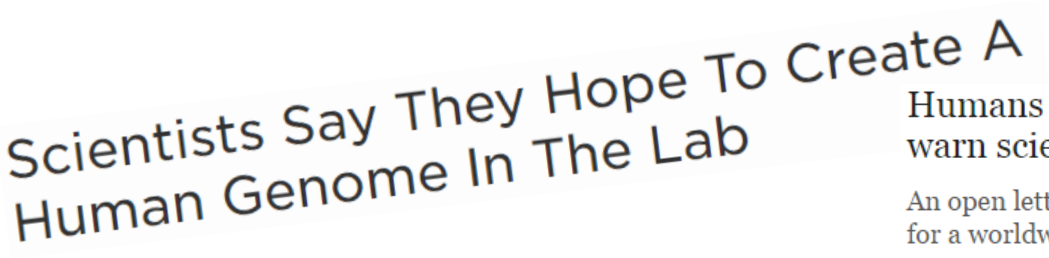
The importance of being: genomic literate
genomic informed

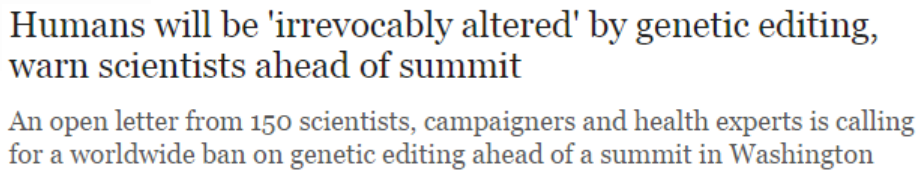
# Health Literacy

# Orphan Black

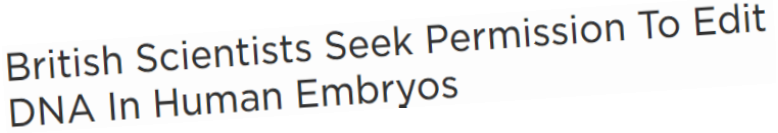
Orphan Black video

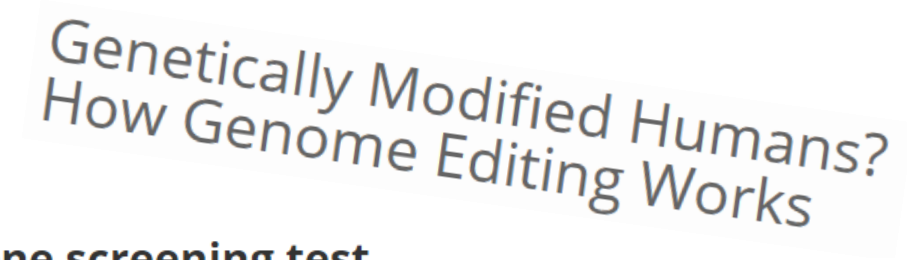
# News Headlines






# Health professional literacy

- Most physicians have no formal training in genetics
- Little research has focused on their understanding of the recent developments in genomics
- Patients ill-informed, inflated expectations, little scientific evidence regarding clinical utility of genomic interventions
- Health professionals have not always kept up to date with the genetic advances
- Need to improve medical education and beyond initial training

PMID 19341496 http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2664958/

# Leading causes of death

1. Heart disease: 614,348
2. Cancer: 591,699
3. Chronic lower respiratory diseases: 147,101
4. Accidents (unintentional injuries): 136,053
5. Stroke (cerebrovascular diseases): 133,103
6. Alzheimer's disease: 93,541
7. Diabetes: 76,488
8. Influenza and pneumonia: 55,227
9. Nephritis, nephrotic syndrome, and nephrosis: 48,146
10. Intentional self-harm (suicide): 42,773

# Definitions

- Genomic Health Literacy
  - The capacity to obtain, process, understand, and use genomic information for health related decision making.
- Genomic Science Literacy
  - The knowledge of basic genetics and genomics concepts and processes needed to build conceptual understanding, and the necessary mathematical knowledge to support this comprehension.

# Genomic literacy

- Genomic health literacy
- Genomic science literacy
- Role of media in genomic literacy

# Genomic Health Literacy

- Lack biology basics
- Lack mathematical concepts
- Low health literacy

# Genomic Science Literacy

- K-12 education unable to keep up with scientific advancements
- Low emphasis on genomics
- Some teachers have misconceptions about genetics/genomics and little understanding
- Teachers need updated skills and have little access to genetic/genomic quality science curriculum
- Encourage partnerships with scientists

# Media role in genomic literacy

- Scientists lack training when communicating with media and general public
- Popular and mass media lack knowledge and often relay incorrect information
- Educators and researchers need to adapt and learn to inform through newer media platforms such as social media and podcasts

**HHS Public Access**

Author manuscript

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Published in final edited form as:

*Genet Med*. 2013 August ; 15(8): 658–663. doi:10.1038/gim.2013.14.

# What Does it Mean to be Genomically Literate? National Human Genome Research Institute Meeting Report

**Belen Hurle, PhD**[1], **Toby Citrin, JD**[2,*], **Jean F. Jenkins, PhD**[3], **Kimberly A. Kaphingst, ScD**[4,*], **Neil Lamb, PhD**[5,*], **Jo Ellen Roseman, PhD**[6,*], and **Vence L. Bonham, JD**[1]

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[3]Genomic Healthcare Branch, National Human Genome Research Institute, National Institutes of Health (NIH), 9000 Rockville Pike, Bethesda, MD 20892-2152, USA

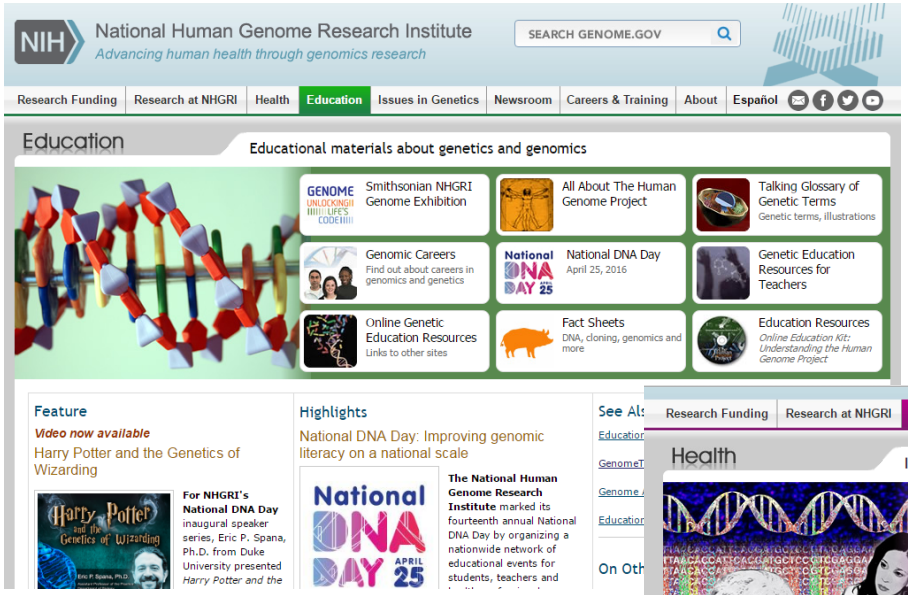
[4]Washington University School of Medicine, Division of Public Health Sciences, Department of

PMID: 23448722

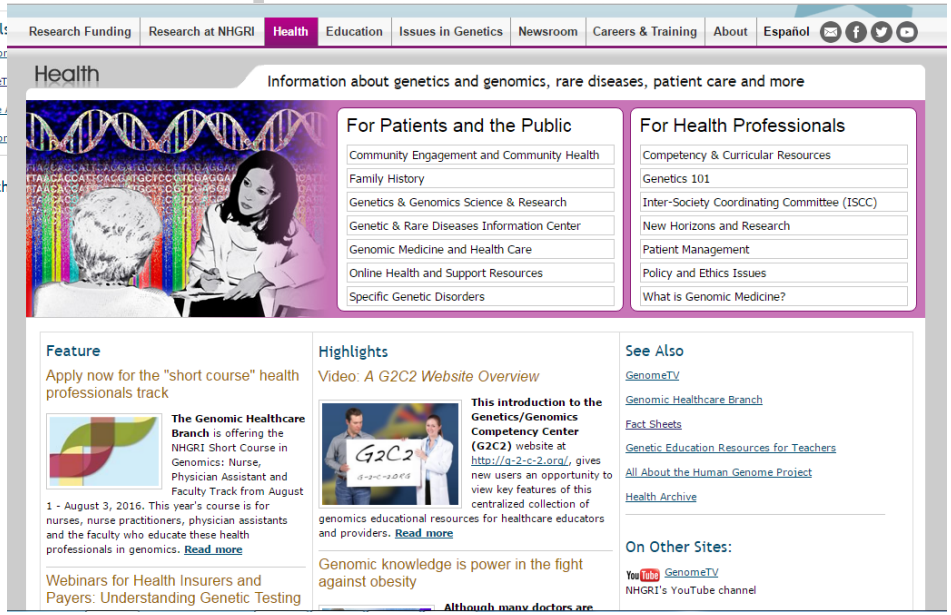
http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4115323/

Genomic discoveries will increasingly advance the science of medicine. Limited genomic literacy may adversely impact the public's understanding and use of the power of genetics and genomics in health care and public health. In November 2011, a meeting was held by the National Human Genome Research Institute to examine the challenge of achieving genomic literacy for the general public, from K-12 to adult education. The role of the media in disseminating scientific messages and in perpetuating, or reducing, misconceptions was also discussed. Workshop participants agreed that genomic literacy will only be achieved through active engagement between genomics experts and the varied constituencies that comprise the public.

# NIH National Human Genome Research Institute



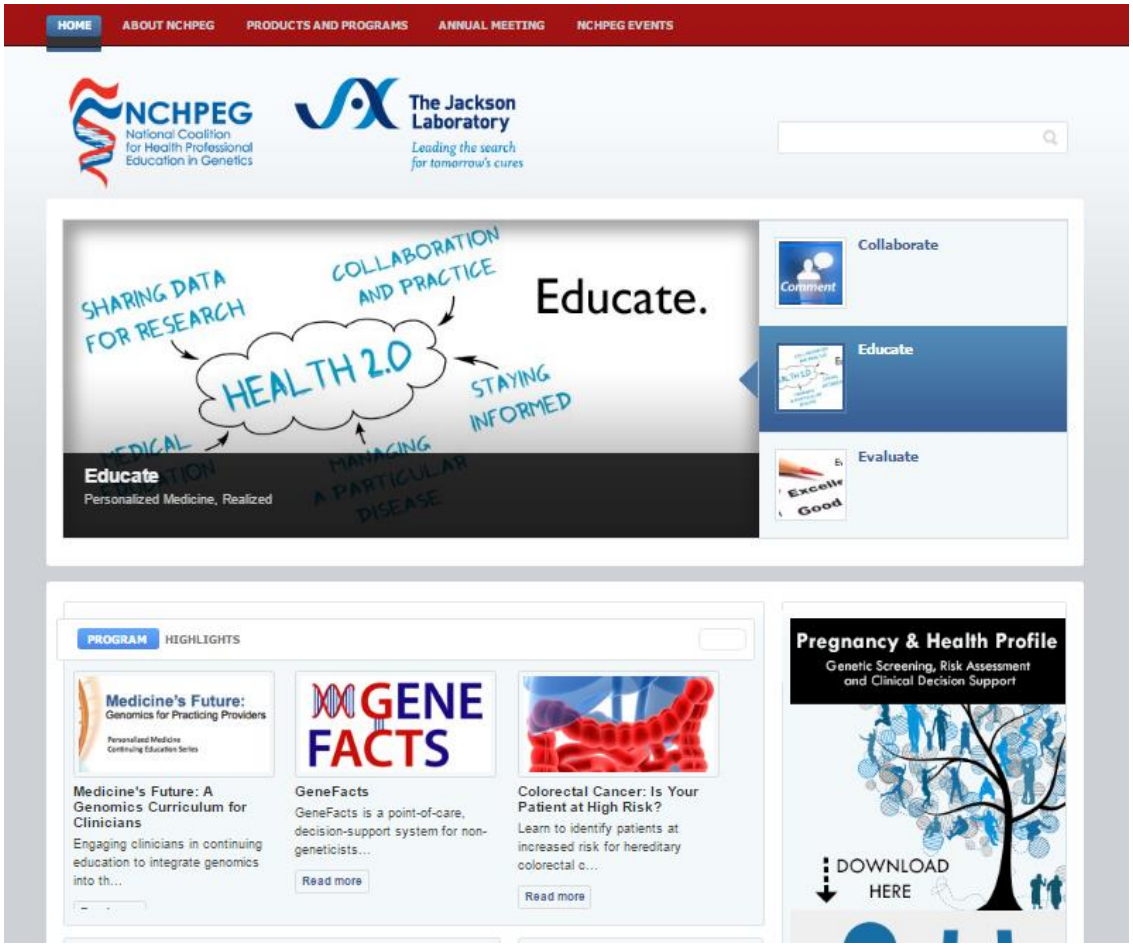






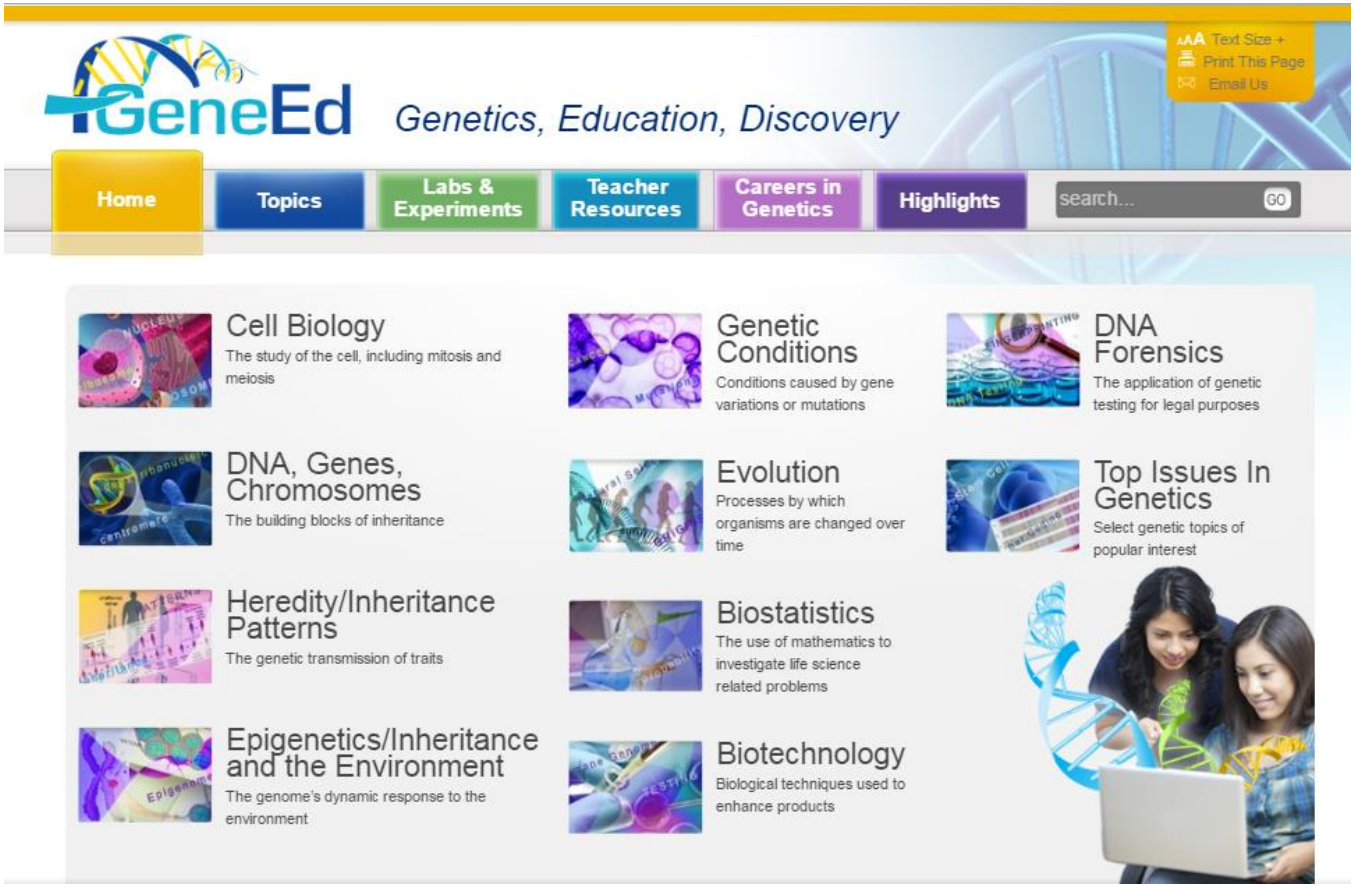
G2C2 http://g-2-c-2.org//

# National Coalition for Health Professional Education in Genetics (NCHPEG)




NCHPEG http://www.nchpeg.org/

# NLM GeneEd

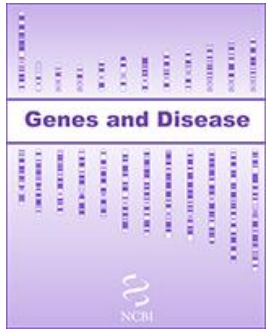



GeneEd https://geneed.nlm.nih.gov/

# NCBI Bookshelf

**Genes and Diseases**

- Organized by the parts of the body that genetic disorders affect
- Over 80 genetic disorder summaries
- Images and interesting facts
- PDF downloads of chapters
- Links to related research literature and pertinent websites




Genes and Diseases

http://www.ncbi.nlm.nih.gov/books/NBK22183/

# Literacy/Education Resources

CDC Centers for Disease Control and Prevention
CDC 24/7: Saving Lives, Protecting People™

Public Health Genomics

ACMG
Translating Genes Into Health®

pgEd Personal Genetics Education Project

ASHG THE AMERICAN SOCIETY OF HUMAN GENETICS
discover. educate. advocate.

GENETIC LITERACY PROJECT
SCIENCE TRUMPS IDEOLOGY

Genes in Life

GENOME UNLOCKING LIFE'S CODE

# Tips for communicating to patients

- Listen, pay attention, respond
- Use plain language
- Use patient's words
- Slow down
- Limit and repeat content
- Show examples
- Invited patient participating
- Use teach back

Health Literacy and Precision Medicine
http://www.nationalacademies.org/hmd/Activities/PublicHealth/HealthLiteracy/2016-MAR-2.aspx

# Tips for communicating to healthcare professionals

- Bring a family member or a friend
- Write down or record the information
- Speak your mind
- Repeat the information as you understand it
- Ask questions (who, what, where, why, how)
- Ask for more information (website, printed handout, a library)

# MedlinePlus Magazine
# My Family Health Portrait

## My Family Health Portrait, A tool from the Surgeon General

Past Issues / Winter 2010 Table of Contents

***My Family Health Portrait*** is an Internet-based tool that makes it easy to create your family's health history. It is simple to fill out. It is private. It is valuable health information that you can share with family members, for their benefit, and with your healthcare practitioner, for your better health.

***My Family Health History*** is available at:
https://familyhistory.hhs.gov/fhh-web/home.action

***Using My Family Health Portrait***, you can:

- Record your family's health history
- Print out and share the history with your family and your healthcare provider
- Save and regularly update your family health history for future use

### Why is it important to know my family medical history?

Your family medical history is a record of health information about you and three generations of close relatives. Family history can be an important risk factor for problems like heart disease, stroke, diabetes, and cancer. A risk factor is anything that increases your chance of getting a disease. The reason a family history can help predict risk is that families share their genes, as well as other factors that affect health, like environment, lifestyles, and habits. A family medical history allows you to take steps to reduce your risk.

### To Find Out More

**MedlinePlus: Family History**
www.nlm.nih.gov/medlineplus/familyhistory.html

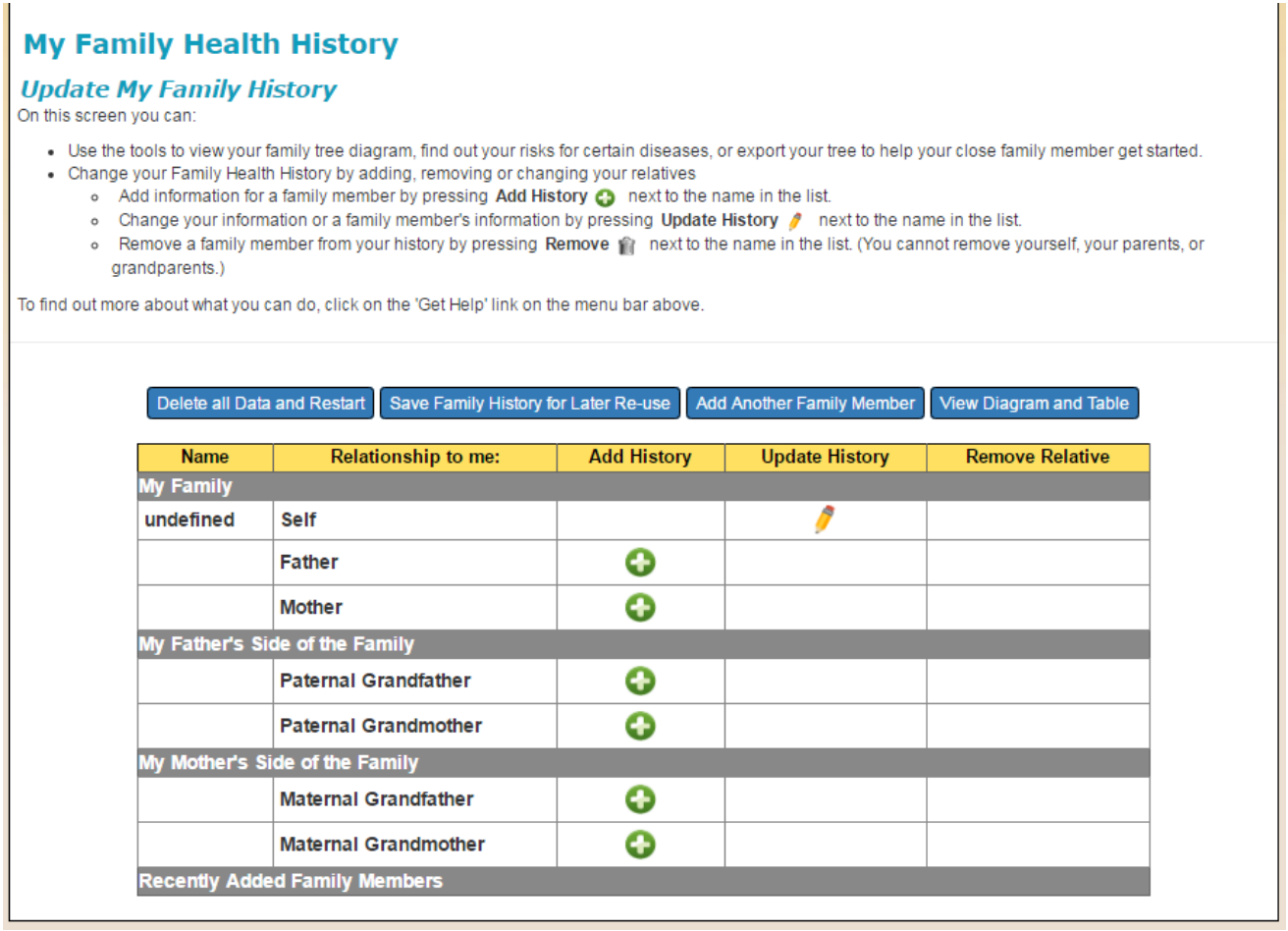
**Family Health History**
www.cdc.gov/genomics/famhistory/index.htm

MedlinePlus Magazine
https://www.nlm.nih.gov/medlineplus/magazine/issues/winter10/articles/winter10pg4.html

# My Family Health Portrait


NHGRI My Family Health Portrait https://www.genome.gov/27527640/family-history-my-family-health-portrait/

# My Family Health History



## My Family Health History

### *Update My Family History*

On this screen you can:

- Use the tools to view your family tree diagram, find out your risks for certain diseases, or export your tree to help your close family member get started.
- Change your Family Health History by adding, removing or changing your relatives
  - Add information for a family member by pressing **Add History** next to the name in the list.
  - Change your information or a family member's information by pressing **Update History** next to the name in the list.
  - Remove a family member from your history by pressing **Remove** next to the name in the list. (You cannot remove yourself, your parents, or grandparents.)

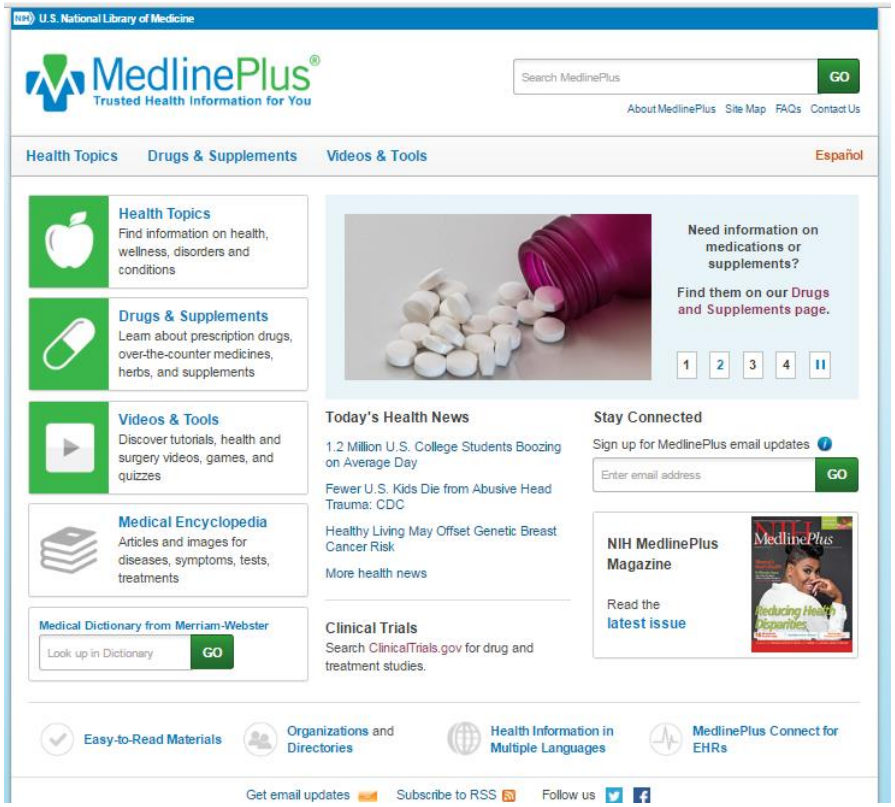
To find out more about what you can do, click on the 'Get Help' link on the menu bar above.

Delete all Data and Restart | Save Family History for Later Re-use | Add Another Family Member | View Diagram and Table

| Name | Relationship to me: | Add History | Update History | Remove Relative |
|---|---|---|---|---|
| **My Family** | | | | |
| undefined | Self | | | |
| | Father | | | |
| | Mother | | | |
| **My Father's Side of the Family** | | | | |
| | Paternal Grandfather | | | |
| | Paternal Grandmother | | | |
| **My Mother's Side of the Family** | | | | |
| | Maternal Grandfather | | | |
| | Maternal Grandmother | | | |
| **Recently Added Family Members** | | | | |

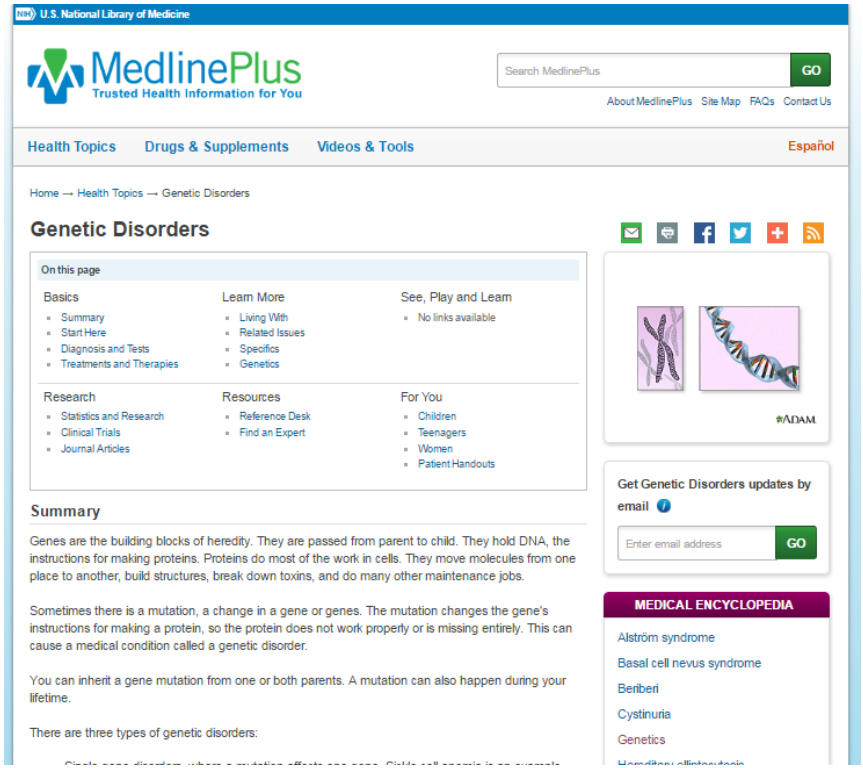
# Consumer Websites

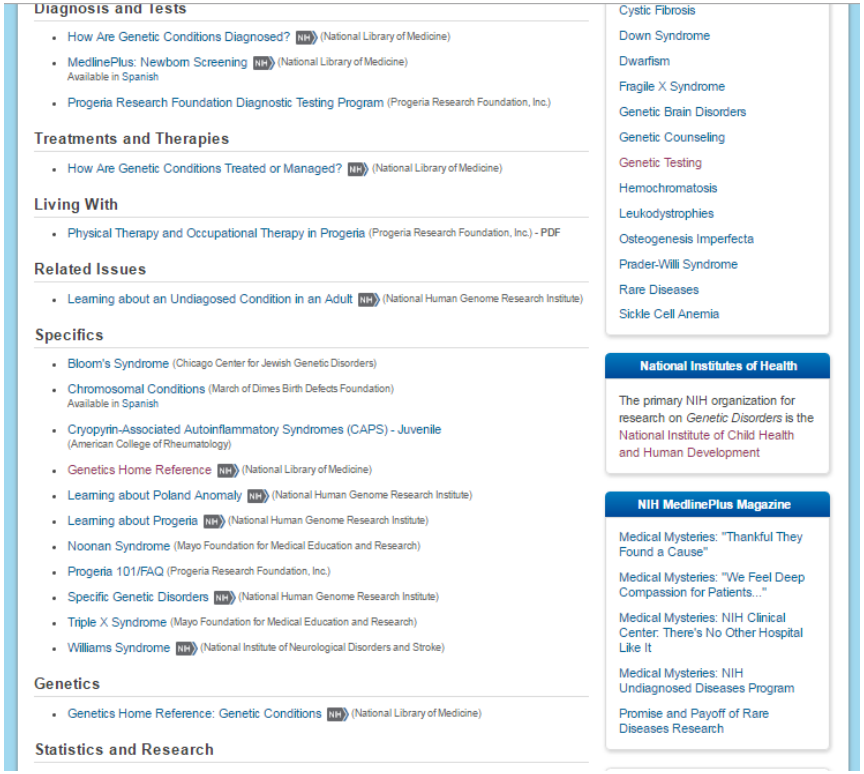
# MedlinePlus




- Health Topic pages:
  - Genetics
  - Genetic testing
  - Genetic counseling
  - Genetic disorders
  - Genes and therapy
- text word search

# MedlinePlus



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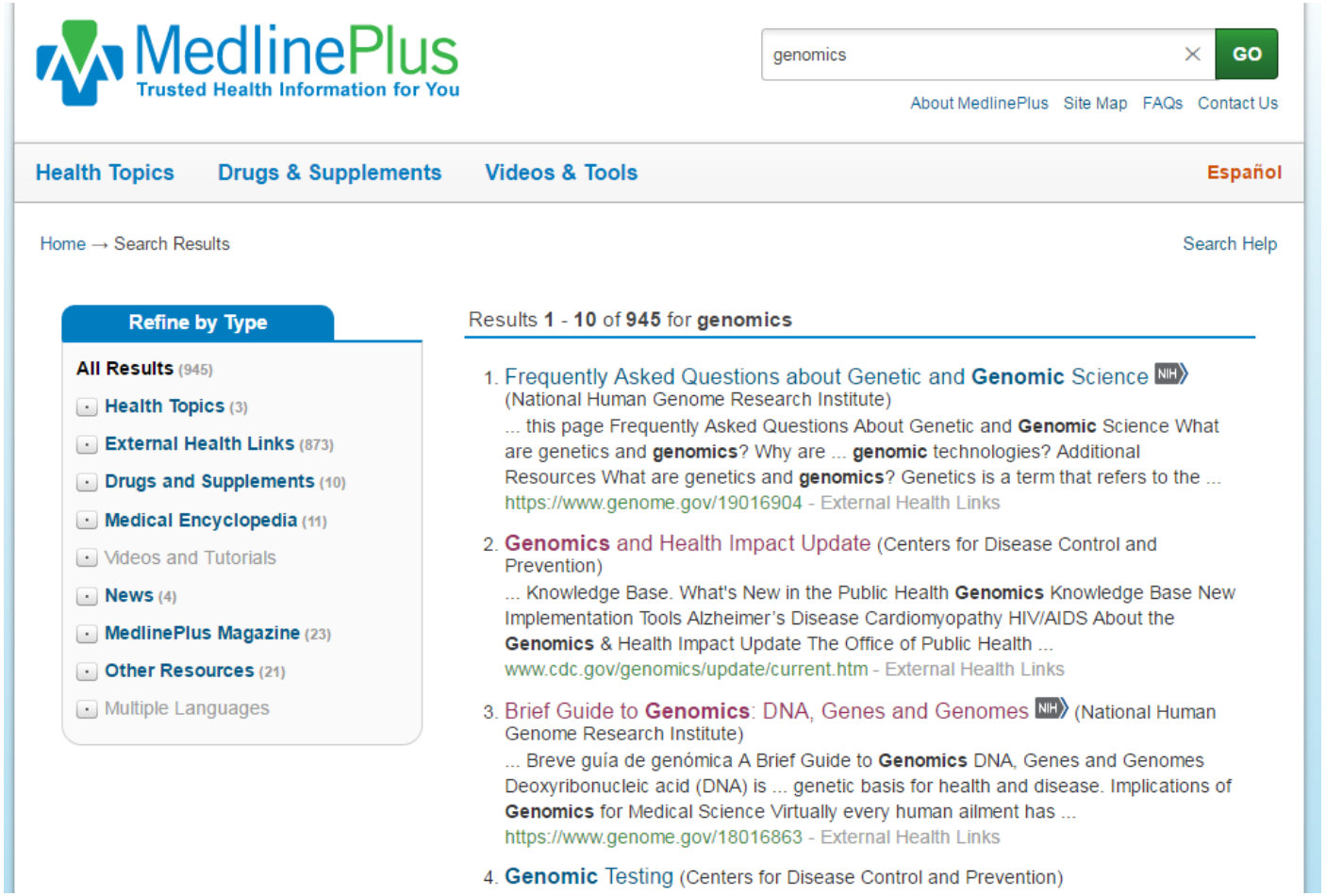
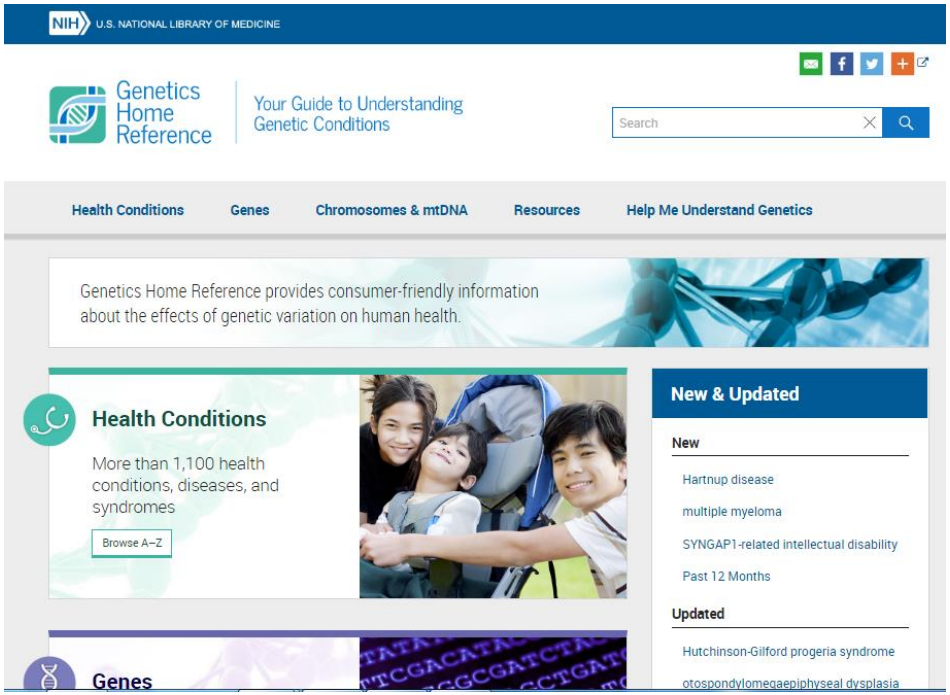


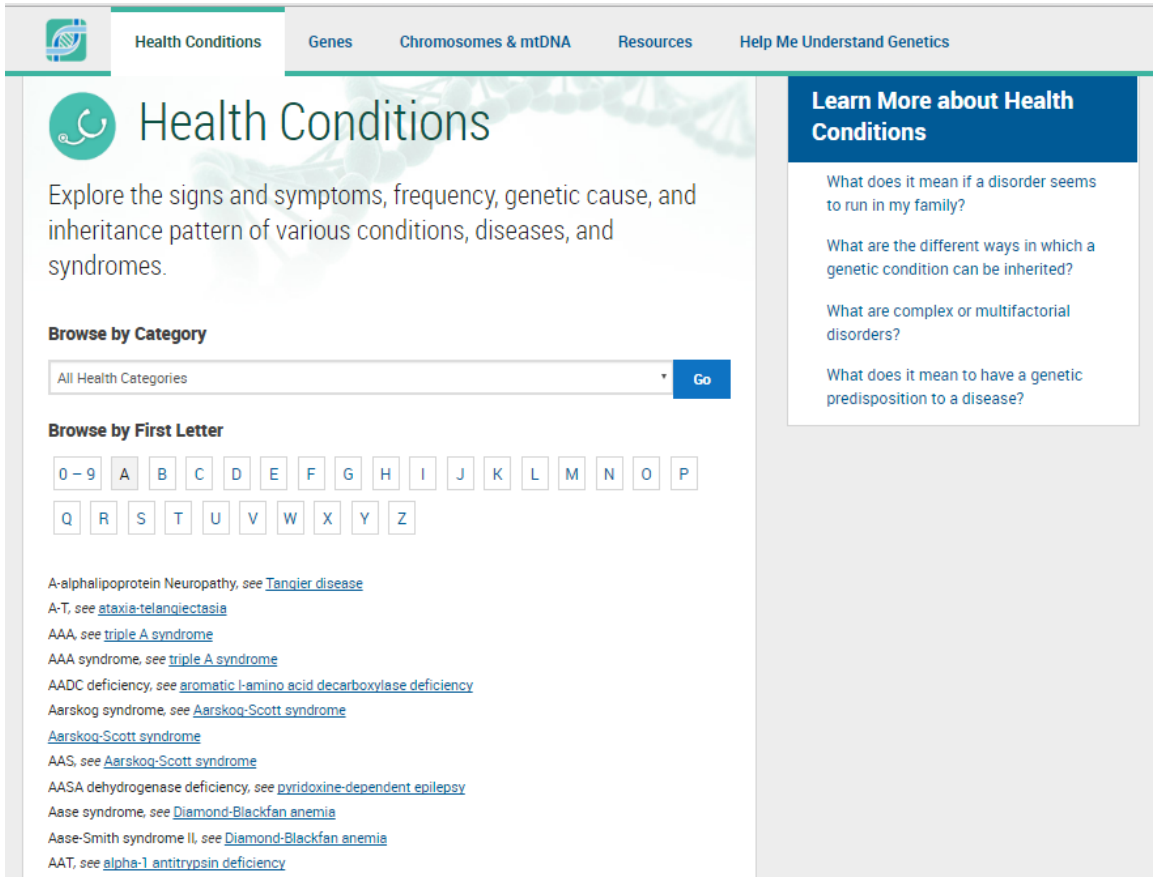
# Genetics Home Reference




- Health conditions
- Genes
- Chromosomes and DNA
- Resources
- Genetic handbook

Genetics Home Reference https://ghr.nlm.nih.gov/

# Genetics Home Reference



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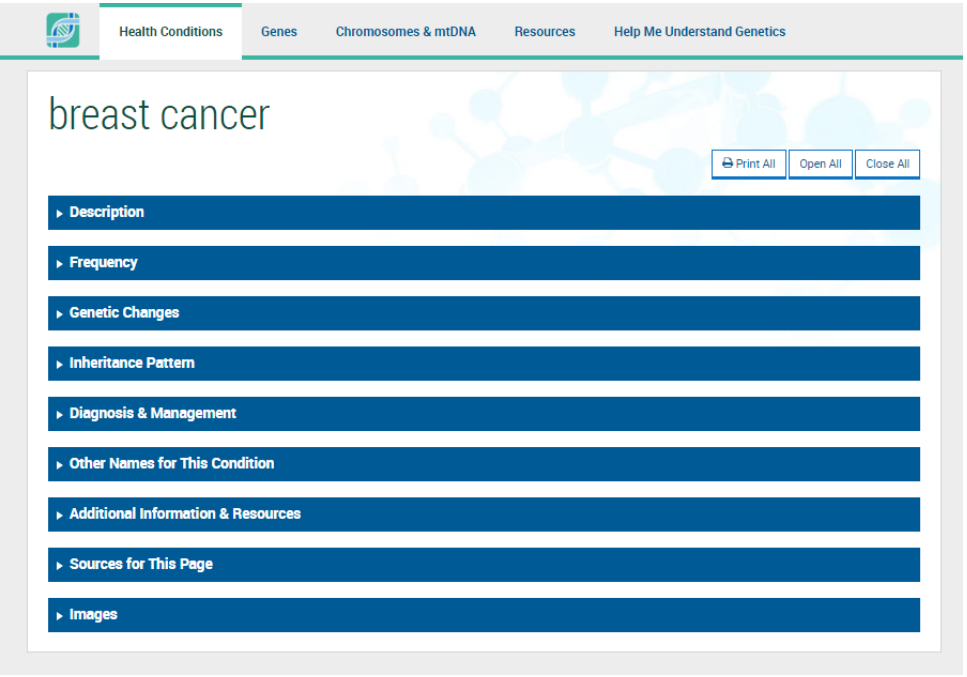


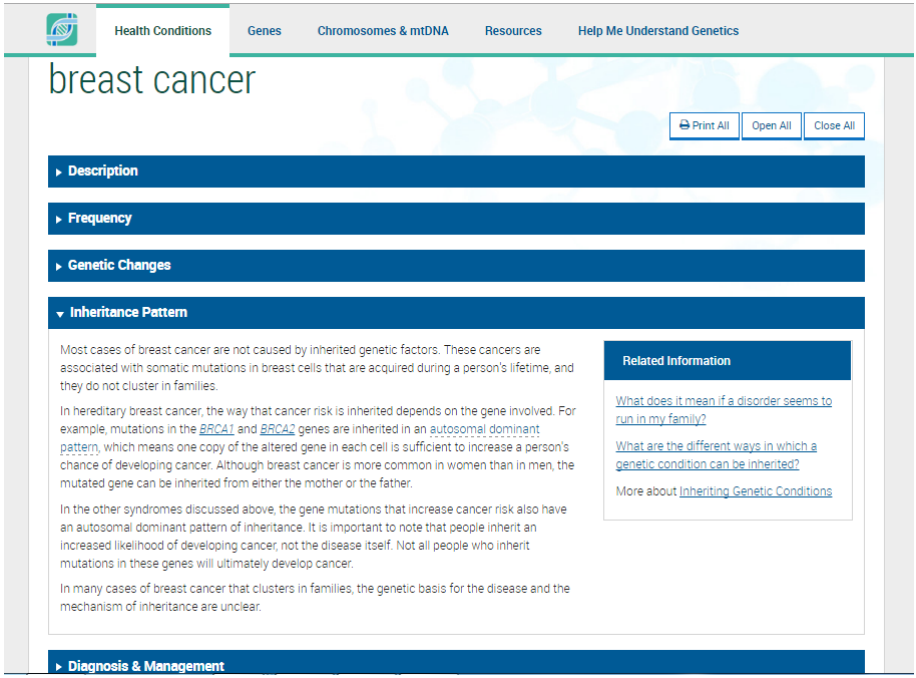

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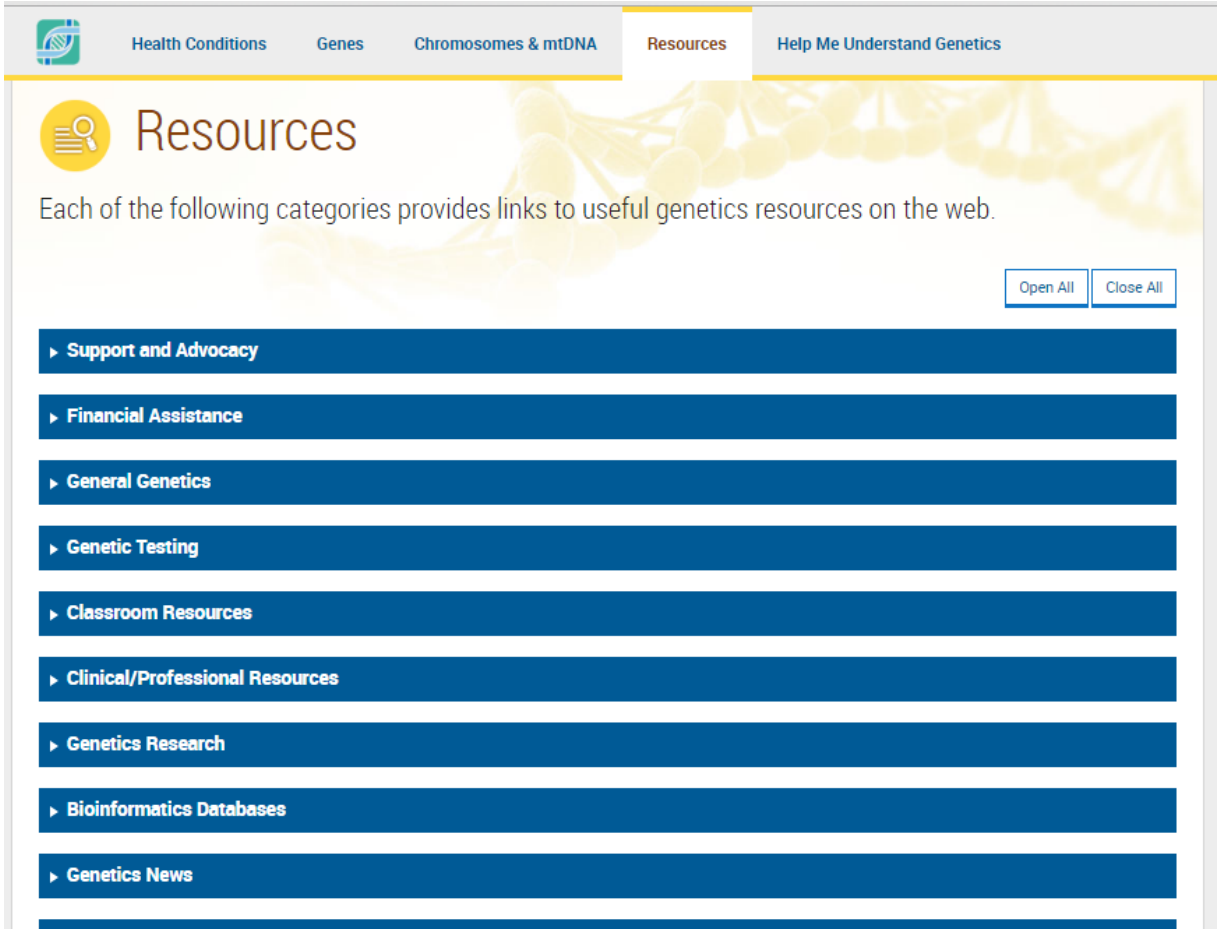

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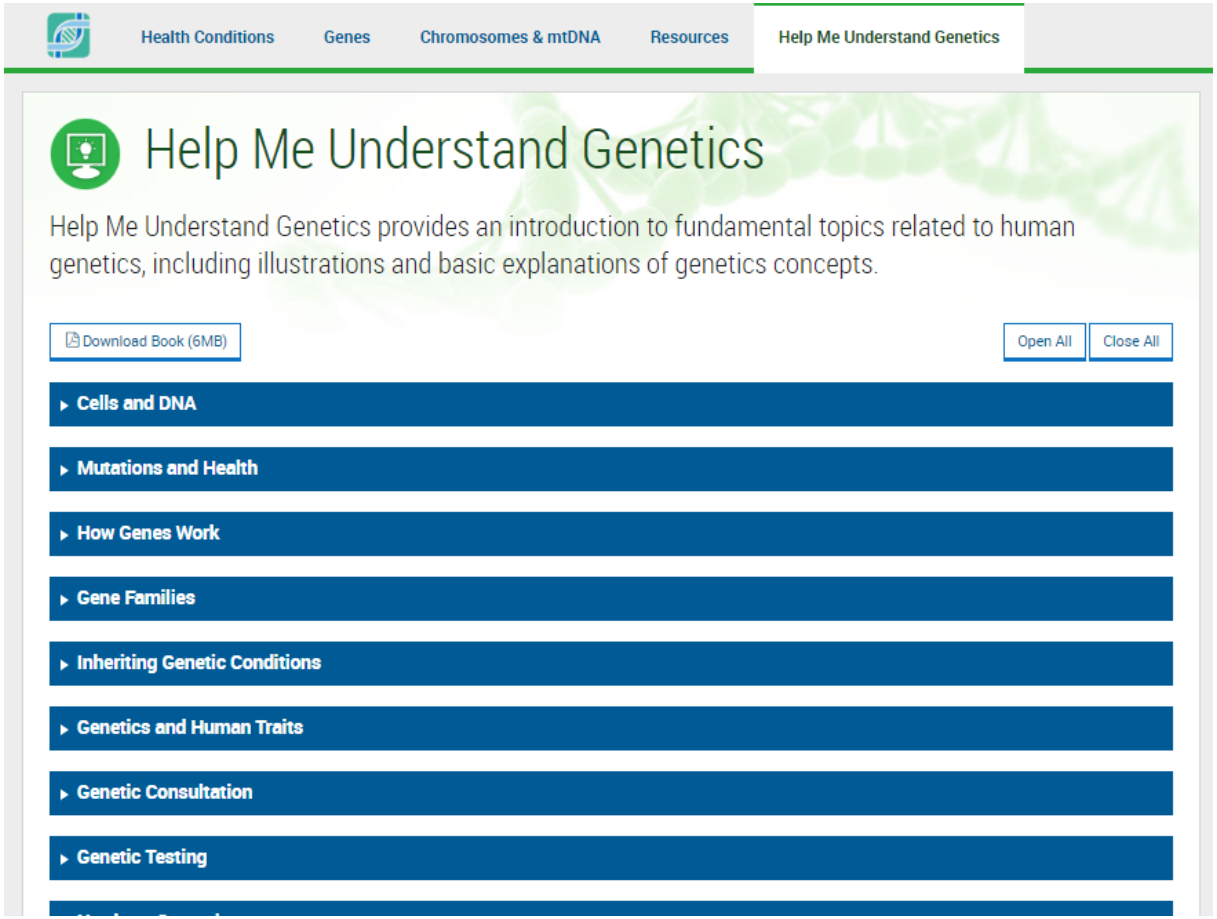

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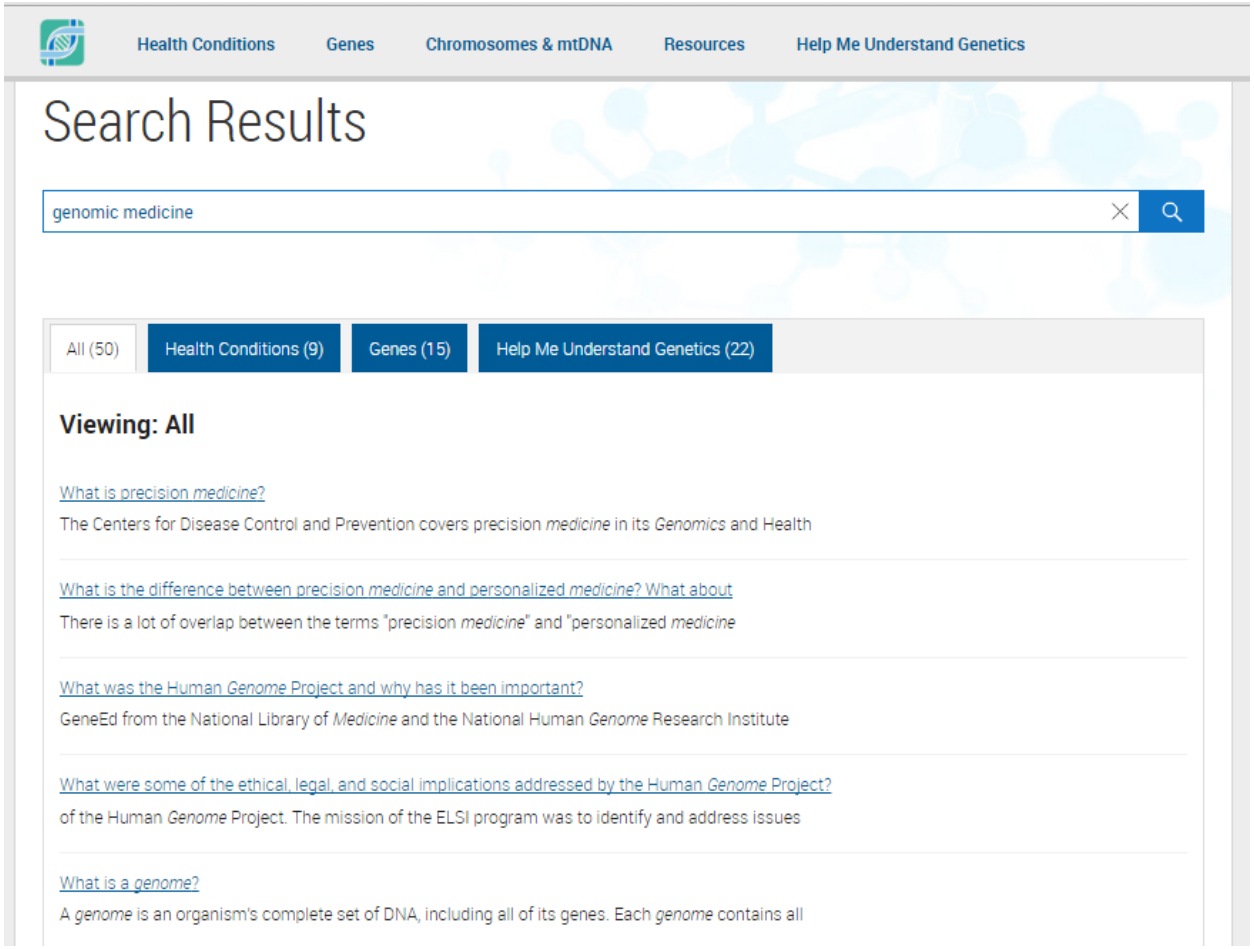
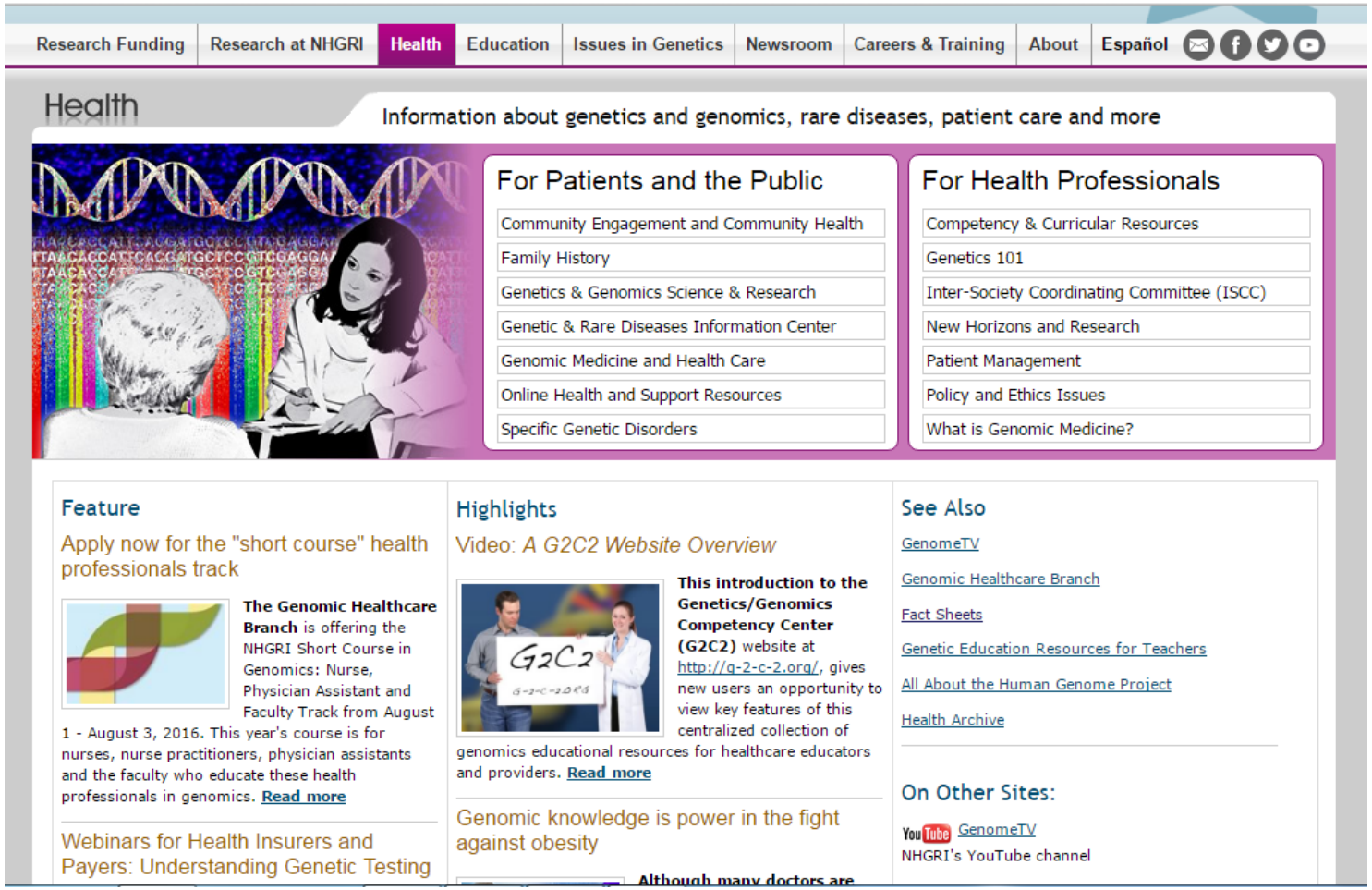


# NIH National Human Genome Research Institute




NHGRI https://www.genome.gov/

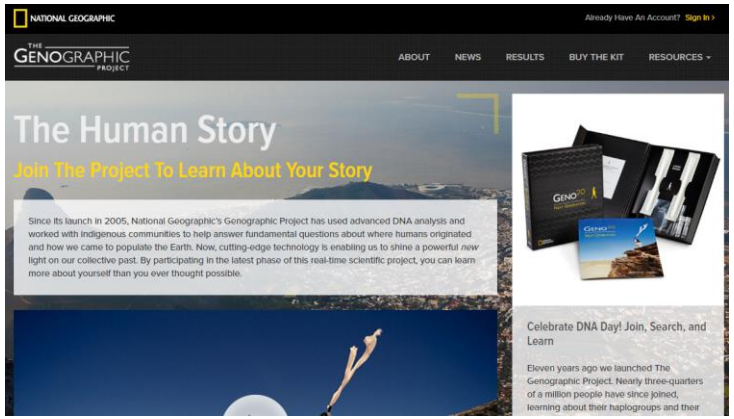
# Direct to Consumer Testing

Genetic Testing

# Direct to Consumer Testing


FUTURA GENETICS

GENE BY GENE



# Concerns

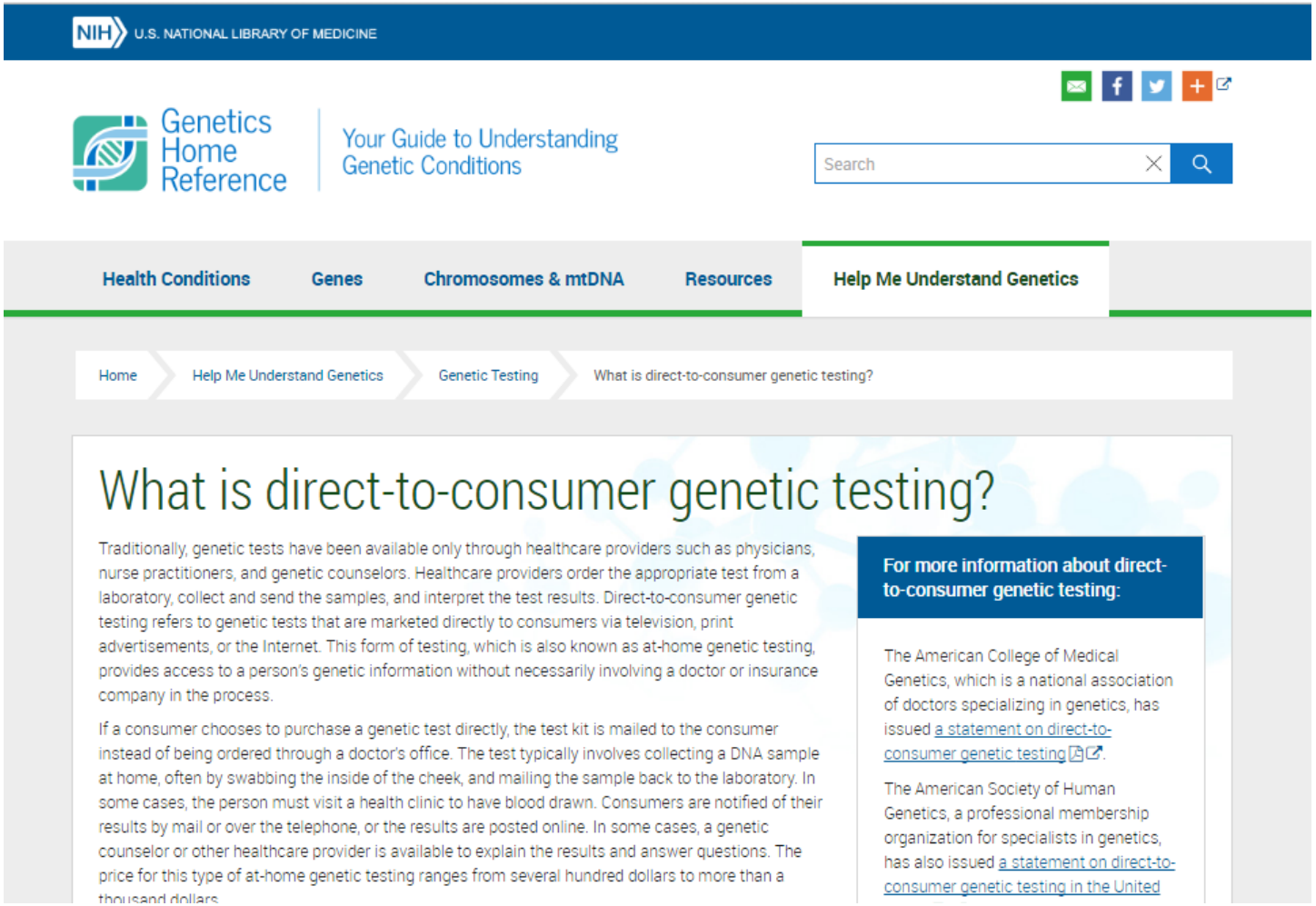
- Privacy
- Legality
- Who has access?
- How useful now?
- What all is being done now and in the future with the information?
- Unexpected surprises?
- Test results can vary among companies
- Validity of tests
- No counseling provided
- Who can get the testing?

# Benefits

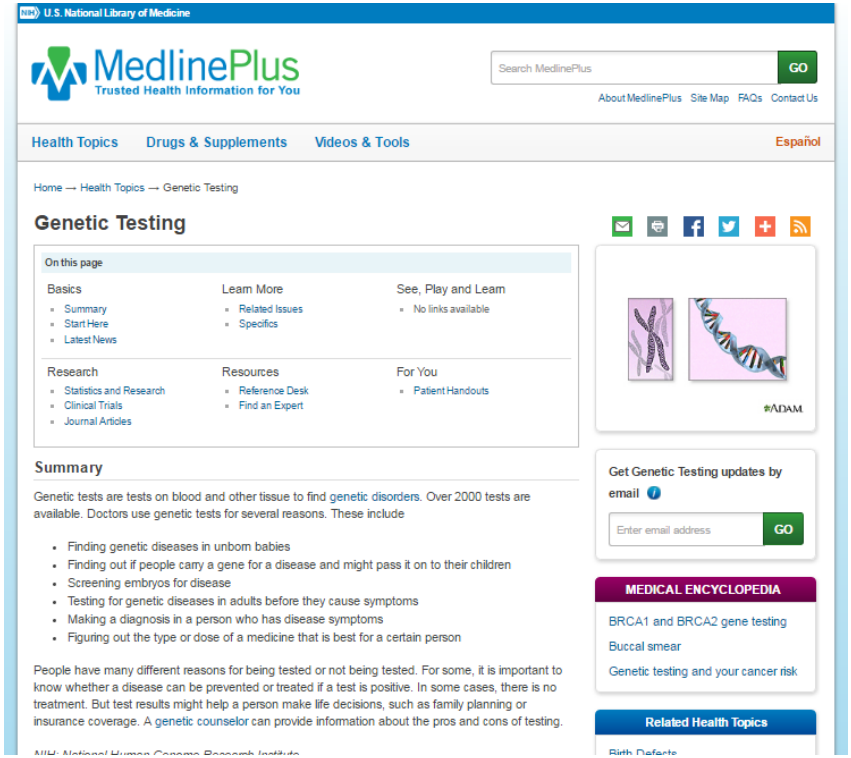
- Learn more about own health
- More effective medical treatments
- Learn more about ethnicity and family history
- Bring awareness to family health issues for future generations
- Motivation to work on health habits
- Encourages patient engagement
- Contributing to advancement of healthcare and science
- Moral obligation

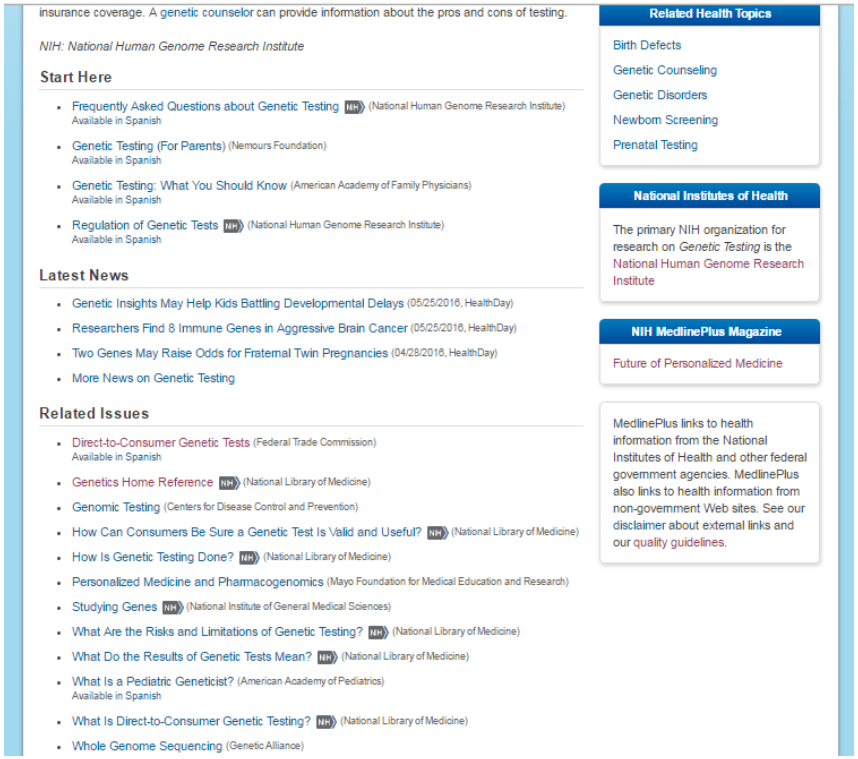

Example of a 23andMe test result

# Genetics Home Reference



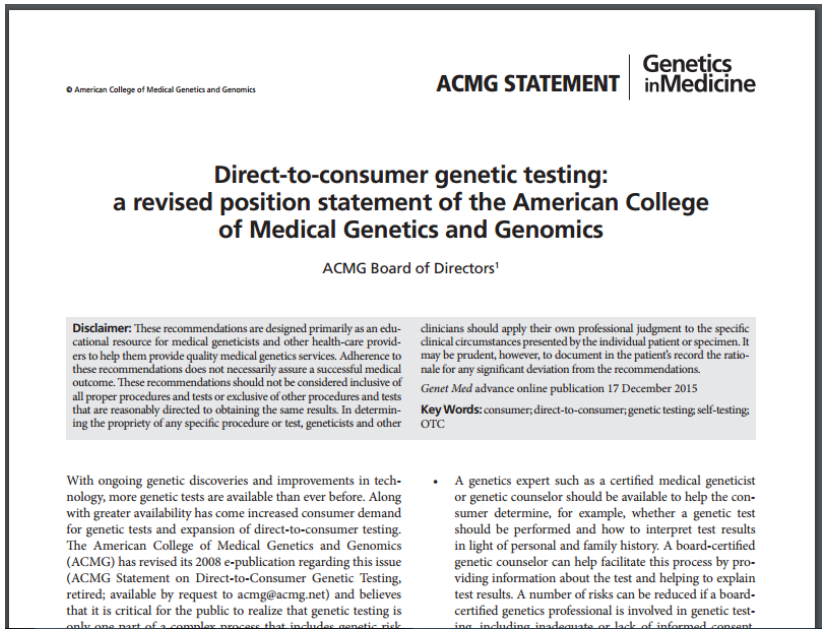
# MedlinePlus



# American College of Medical Genetics and Genomics

| | | |
|---|---|---|
| **Folic Acid** | | |
| Folic Acid and Neural Tube Defects | 2010, ACMG | 2010 Genet Med 13:6:593-596 |
| **Genetic Discrimination** | | |
| Points to Consider in Preventing Unfair Discrimination Based on Genetic Disease Risk: A Position Statement of the American College of Medical Genetics and Genomics | 2001, ACMG Reaffirmed 2005 | 2001 Genet Med 3:6: 436-437 |
| **Genetic Services** | | |
| Clinical utility of genetic and genomic services: a position statement of the American College of Medical Genetics and Genomics | 2015, ACMG | 2015 Genet Med 7:6:505-507 |
| **Genetic Testing** | | |
| ACMG Revised Position Statement on Direct-to-Consumer Genetic Testing | 2015, ACMG | 2016 Genet Med 18:2:207-208 |
| ACMG position statement on prenatal/ preconception expanded carrier screening | 2013, ACMG | 2013 Genet Med 15:6:482-483 |
| Technical report: ethical and policy issues in genetic testing and screening of children | 2013, ACMG/AAP | 2013 Genet Med 15:3:234-245 |
| Risk categorization for oversight of laboratory-developed tests for inherited conditions | 2013, ACMG | 2013 Genet Med 15:4:314-5 |
| Genetic Testing in Adoption (ACMG/ASHG) | 2000, ASHG | 2000 Am J Hum Genet 66:761-767 |
| **Genomic Sequencing** | | |
| ACMG Policy Statement: Updated Recommendations Regarding Analysis and Reporting of Secondary Findings in Clinical Genome-Scale Sequencing | 2015, ACMG | 2015 Genet Med 17:1:68-69 |
| ACMG Recommendations for reporting of incidental findings in clinical exome and | 2013, | 2013 Genet |



© American College of Medical Genetics and Genomics

ACMG STATEMENT | Genetics inMedicine

**Direct-to-consumer genetic testing: a revised position statement of the American College of Medical Genetics and Genomics**

ACMG Board of Directors[1]

**Disclaimer:** These recommendations are designed primarily as an educational resource for medical geneticists and other health-care providers to help them provide quality medical genetics services. Adherence to these recommendations does not necessarily assure a successful medical outcome. These recommendations should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, geneticists and other clinicians should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from the recommendations.

*Genet Med* advance online publication 17 December 2015

**Key Words:** consumer; direct-to-consumer; genetic testing; self-testing; OTC

With ongoing genetic discoveries and improvements in technology, more genetic tests are available than ever before. Along with greater availability has come increased consumer demand for genetic tests and expansion of direct-to-consumer testing. The American College of Medical Genetics and Genomics (ACMG) has revised its 2008 e-publication regarding this issue (ACMG Statement on Direct-to-Consumer Genetic Testing, retired; available by request to acmg@acmg.net) and believes that it is critical for the public to realize that genetic testing is

- A genetics expert such as a certified medical geneticist or genetic counselor should be available to help the consumer determine, for example, whether a genetic test should be performed and how to interpret test results in light of personal and family history. A board-certified genetic counselor can help facilitate this process by providing information about the test and helping to explain test results. A number of risks can be reduced if a board-certified genetics professional is involved in genetic testing, including inadequate or lack of informed consent

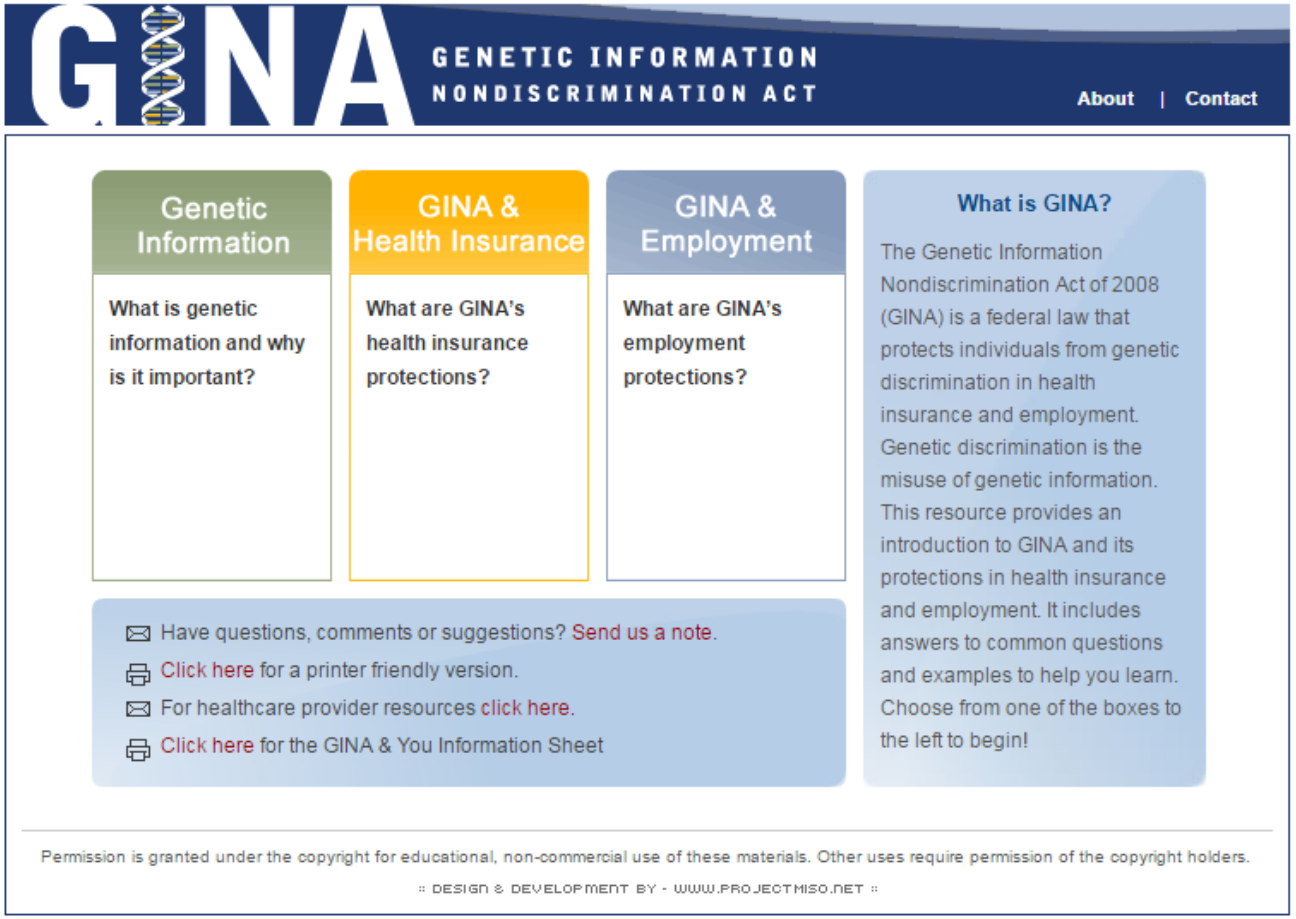
ACMG https://www.acmg.net/

# Ethics and Privacy

# Societal Concerns

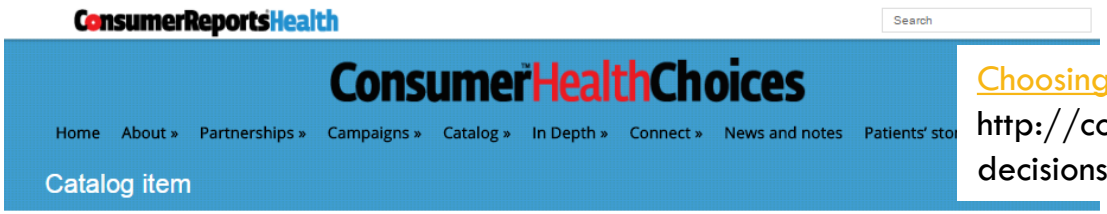
- Who should have access to personal genetic information, and how will it be used?
- Who owns and controls genetic information?
- How does personal genetic information affect an individual and society's perceptions of that individual?
- What are the larger societal issues raised by new reproductive technologies?
- How will genetic tests be evaluated and regulated for accuracy, reliability and utility?
- How do we prepare healthcare professionals and the public?
- What is considered acceptable diversity?
- Where is the line between medical treatment and enhancement?
- Should testing be performed when no treatment is available?

# GINA



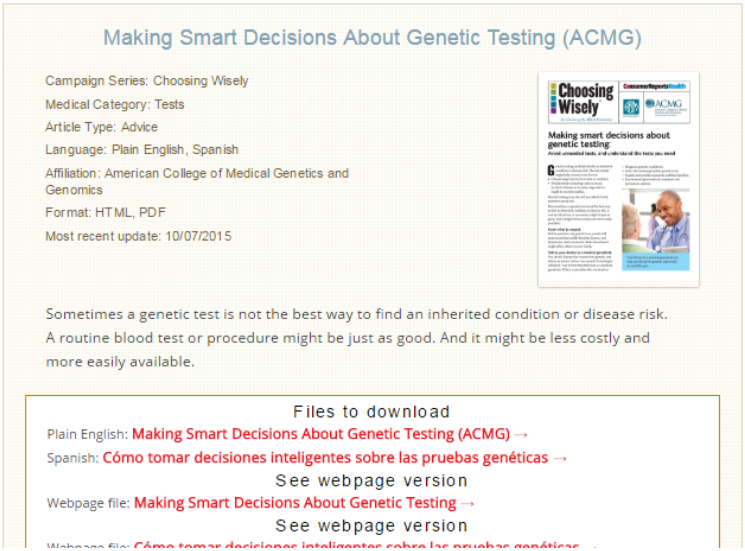

GINA Help http://ginahelp.org/

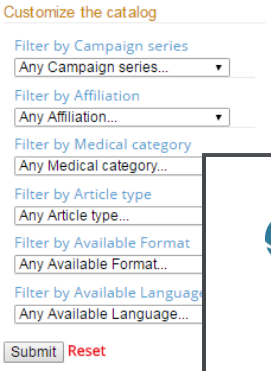
# Choosing Wisely




Choosing Wisely

http://consumerhealthchoices.org/catalog/making-smart-decisions-about-genetic-testing-acmg/



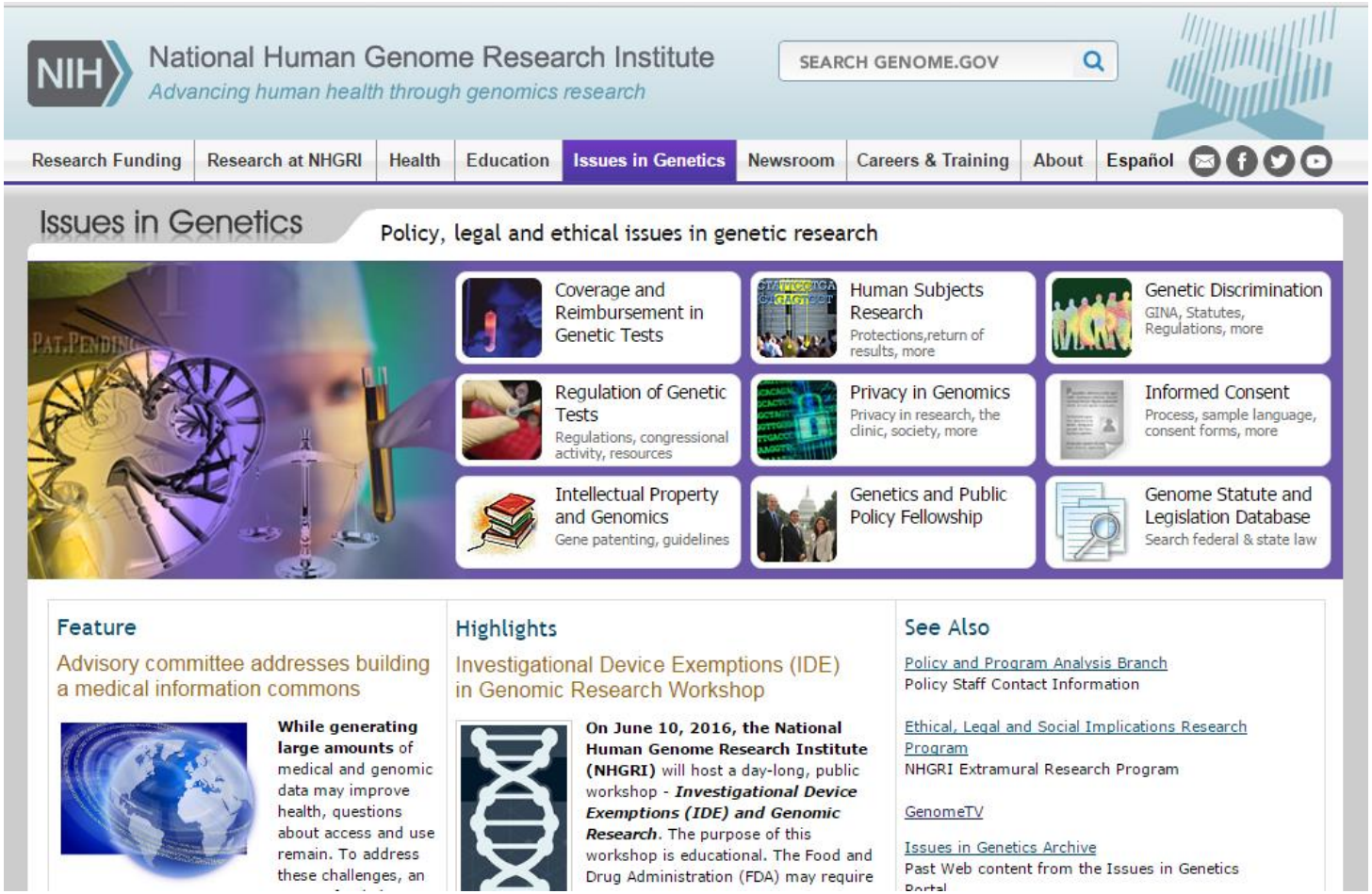





ACMG

https://www.acmg.net/docs/ACMG_Choosing Wisely_Final.pdf

# NIH National Human Genome Research Institute



# American Academy of Pediatrics
# American College of Medical Genetics and Genomics

Pediatrics
March 2013, VOLUME 131 / ISSUE 3
From the American Academy of Pediatrics
Policy Statement

AAP

http://pediatrics.aappublications.org/content/131/3/620

## Ethical and Policy Issues in Genetic Testing and Screening of Children

COMMITTEE ON BIOETHICS, COMMITTEE ON GENETICS, AND, THE AMERICAN COLLEGE OF MEDICAL GENETICS AND, GENOMICS SOCIAL, ETHICAL, AND LEGAL ISSUES COMMITTEE

Article | Info & Metrics | Comments

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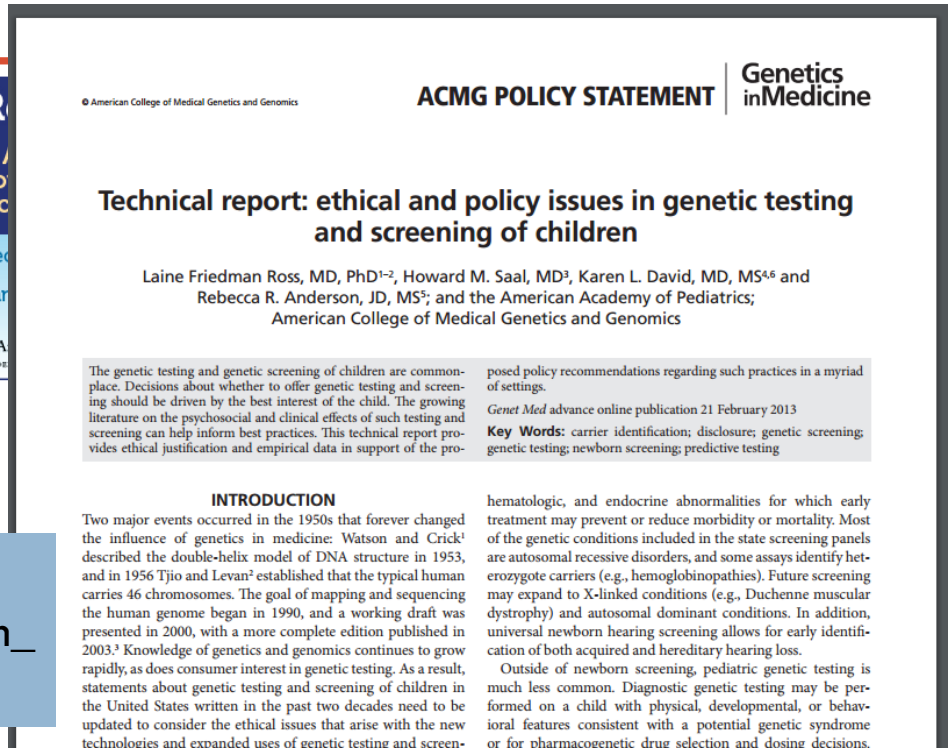
### Abstract

The genetic testing and genetic screening of children are commonplace. Decisions about whether to offer genetic testing and screening should be driven by the best interest of the child. The growing literature on the psychosocial and clinical effects of such testing and screening can help inform best practices. This policy statement represents recommendations developed collaboratively by the American Academy of Pediatrics and the American College of Medical Genetics and Genomics with respect to many of the scenarios in which genetic testing and screening can occur.

ACMG

https://www.acmg.net/docs/genetic_testing_in_children_preprint_gim2012176a.pdf





ACMG POLICY STATEMENT | Genetics inMedicine

## Technical report: ethical and policy issues in genetic testing and screening of children

Laine Friedman Ross, MD, PhD[1–2], Howard M. Saal, MD[3], Karen L. David, MD, MS[4,6] and Rebecca R. Anderson, JD, MS[5]; and the American Academy of Pediatrics; American College of Medical Genetics and Genomics

The genetic testing and genetic screening of children are commonplace. Decisions about whether to offer genetic testing and screening should be driven by the best interest of the child. The growing literature on the psychosocial and clinical effects of such testing and screening can help inform best practices. This technical report provides ethical justification and empirical data in support of the proposed policy recommendations regarding such practices in a myriad of settings.

*Genet Med* advance online publication 21 February 2013

**Key Words:** carrier identification; disclosure; genetic screening; genetic testing; newborn screening; predictive testing

### INTRODUCTION

Two major events occurred in the 1950s that forever changed the influence of genetics in medicine: Watson and Crick[1] described the double-helix model of DNA structure in 1953, and in 1956 Tjio and Levan[2] established that the typical human carries 46 chromosomes. The goal of mapping and sequencing the human genome began in 1990, and a working draft was presented in 2000, with a more complete edition published in 2003.[3] Knowledge of genetics and genomics continues to grow rapidly, as does consumer interest in genetic testing. As a result, statements about genetic testing and screening of children in the United States written in the past two decades need to be updated to consider the ethical issues that arise with the new technologies and expanded uses of genetic testing and screen-

hematologic, and endocrine abnormalities for which early treatment may prevent or reduce morbidity or mortality. Most of the genetic conditions included in the state screening panels are autosomal recessive disorders, and some assays identify heterozygote carriers (e.g., hemoglobinopathies). Future screening may expand to X-linked conditions (e.g., Duchenne muscular dystrophy) and autosomal dominant conditions. In addition, universal newborn hearing screening allows for early identification of both acquired and hereditary hearing loss.

Outside of newborn screening, pediatric genetic testing is much less common. Diagnostic genetic testing may be performed on a child with physical, developmental, or behavioral features consistent with a potential genetic syndrome or for pharmacogenetic drug selection and dosing decisions.

# Informing the Public

CRG Council for Responsible Genetics

Genetic Alliance

# Precision Medicine

"…a bold new research effort to revolutionize how we improve health and treat disease."

PMI announcement https://www.whitehouse.gov/precision-medicine

# Precision Medicine Initiative

**Mission statement:**

*To enable a new era of medicine through research, technology, and policies that empower patients, researchers, and providers to work together toward development of individualized care.*


PMI announcement https://www.whitehouse.gov/precision-medicine

# Precision Medicine is…

- Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.
- Instead of what treatment is right for this disease it is what treatment  is right for this patient.

# Precision Medicine Initiative

- **Near Term goals:**
- Clinical trials focusing on pediatric cancers and drug therapies for adults
- Use of combination therapies
- Overcoming drug resistance
- **Long Term Goals:**
- Create research cohort of 1 million volunteers
- New model of medicine
  - engage participants
  - responsible data sharing
  - privacy protection
- Advance pharmacogenomics
- Identify new targets for treatment and prevention
- Test if mobile devices encourages healthy behaviors
- Lay scientific foundation for many diseases

PMI Infographic http://syndication.nih.gov/multimedia/pmi/infographics/pmi-infographic.pdf

# Precision Medicine Cohort Program

- Two ways to participate

1. Through the cohort website
2. With participating health care provider organization

# NIH and Precision Medicine Initiative

## PRECISION MEDICINE INITIATIVE COHORT PROGRAM

### Precision Medicine Initiative

- Scale and Scope
- Participation
- Program Components
- Funding
- FAQ
- Advisory Groups
- Events
- Announcements
- PMI in the News
- Multimedia


NIH awards $55 million to build million-person precision medicine study

Learn about the key components of the PMI Cohort Program

### About the Precision Medicine Initiative Cohort Program

Far too many diseases do not have a proven means of prevention or effective treatments. We must gain better insights into the biological, environmental, and behavioral influences on these diseases to make a difference for the millions of Americans who suffer from them. Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person. While some advances in precision medicine have been made, the practice is not currently in use for most diseases.

### Email Updates

Sign up to receive email updates about the Precision Medicine Initiative.

Sign up for updates

### Related Links

- PMI Working Group Final Report pdf
- NEJM Perspective: A New Initiative on Precision Medicine
- White House Precision Medicine Web Page
- White House Fact Sheet: President Obama's Precision Medicine Initiative
- Precision Medicine Initiative and Cancer Research
- Precision Medicine Initiative YouTube Channel

PMI Cohort Program ://www.nih.gov/precision-medicine-initiative-cohort-program

# MedlinePlus Magazine- Fall 2015


**Health Care Tailored to You**

# CDC

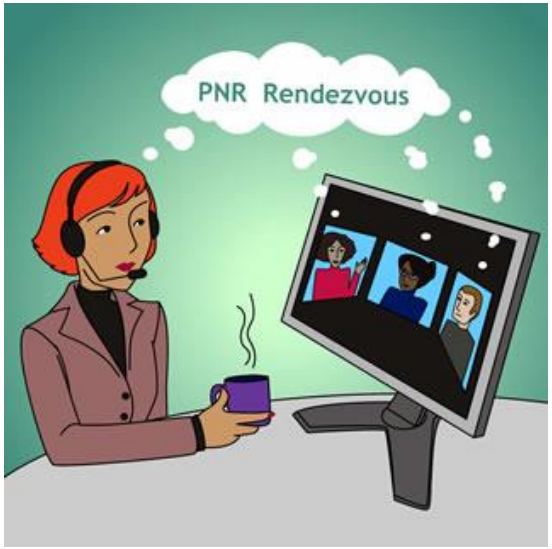

[CDC Public Health Genomics](http://www.cdc.gov/genomics/)

http://www.cdc.gov/genomics/


[Genomics health blog](http://blogs.cdc.gov/genomics/)

http://blogs.cdc.gov/genomics/

# PNR Rendezvous




**Informed Consent and the Precision Medicine Initiative**

September 21, 2016 1PM

Presenter: **Malia Fullerton,**

Associate Professor of Bioethics and Humanities at the University of Washington School of Medicine

# Library role

- “Preparing the public to make educated personal and family health decisions in a time of rapidly evolving genetic and genomic knowledge will require new partnerships between the education system, health care systems, the government, community advocacy organizations, consumers and the media.”

# Show What You Know!

- The 1000 Genomes Project was undertaken in order to increase the _____ of the genomes represented in public databases.
- What term refers to strategies for determining what treatment is right for an INDIVIDUAL rather than what treatment is recommended for a DISEASE?
- Clinicians are not concerned about <u>all</u> genetic variants – only those that are _____.
- True or False? GINA (Genetic Information Nondiscrimination Act) protects you from life insurance discrimination.
- True or False? A genetic variant may originally be classified as "likely pathogenic" and later classified as "likely benign."
- What resource would you recommend to consumers who wanted to learn more about a genetic condition?
- What is a good starting place for finding genetic information for clinicians?

# Questions?

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**Presentation resources**

https://nnlm.gov/pnr/training/presentations